

Claims:

1. A method for the diagnosis of a polymorphism in an EP1-R gene in a human, which method comprises determining the sequence of the nucleic acid of the human at one or more of positions 344, 621-627, 793-799, 908, 1136, 1160, 1189, 1458, 1656, 2448, 2531, 3348, 3432 and 3622 in the EP1-R gene as defined by the positions in SEQ ID NO.1, or the sequence of the amino acid in the EP1-R protein at positions 126 or 154; and determining the status of the human by reference to polymorphism in the EP1-R gene or protein.
- 10 2. A method according to claim 1 wherein the polymorphism is selected from the group in which, according to the position in SEQ ID NO. 1, the polymorphism at position 344 is presence of G and/or A, the polymorphism at position 621-627 is deletion of one or more of the seven Gs, the polymorphism at position 793-799 is deletion of one or more of the seven Gs, the polymorphism at position 908 is presence of C and/or T, the polymorphism at position 1136 is presence of G and/or C, the polymorphism at position 1160 is presence of T and/or C, the polymorphism at position 1189 is presence of G and/or A, the polymorphism at position 1458 is presence of A and/or G, the polymorphism at position 1656 is presence of T and/or G, the polymorphism at position 2448 is presence of T and/or C, the polymorphism at position 2531 is presence of G and/or A, the polymorphism at position 3348 is presence of C and/or T, the polymorphism at position 3432 is presence of C and/or G and, the polymorphism at position 3622 is presence of G and/or A.
- 15 3. A method as claimed in claim 1 or 2, wherein the nucleic acid region containing the potential single nucleotide polymorphism is amplified by polymerase chain reaction prior to determining the sequence.
- 20 4. A method as claimed in any of claims 1 - 3, wherein the presence or absence of the single nucleotide polymorphism is detected by reference to the loss or gain of, optionally engineered, sites recognised by restriction enzymes.

5. A method according to claim 1 or claim 2, in which the sequence is determined by a method selected from ARMS-allele specific amplification, allele specific hybridisation, oligonucleotide ligation assay and restriction fragment length polymorphism (RFLP).

6. A method according to claim 1 wherein the presence of a polymorphic amino acid residue in 5 the EP1-R protein is determined by immunological methods such as enzyme linked immunosorbent assay (ELISA).

7. A method as claimed in any of the preceding claims for use in assessing the predisposition and/or susceptibility of an individual to EP1-R mediated diseases.

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8. A method for the diagnosis of EP1-R-mediated disease, which method comprises:

- i) obtaining sample nucleic acid from an individual,
- ii) detecting the presence or absence of a variant nucleotide at one or more of positions 344, 621-627, 793-799, 908, 1136, 1160, 1189, 1458, 1656, 2448, 2531, 3348, 3432 and 3622 15 in the EP1-R gene as defined by the positions in SEQ ID NO.1; and
- iii) determining the status of the individual by reference to polymorphism in the EP1-R gene.

9. A method for the diagnosis of EP1-R- mediated disease, which method comprises:

- i) obtaining a protein containing sample from an individual;
- ii) detecting the presence or absence of a variant EP1-R polypeptide on the basis of the 20 presence of a polymorphic amino acid at either or both amino acid positions: 126 and 154; and,
- iii) determining the status of the human by reference to the presence or absence of a polymorphism in EP1-R protein.

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10. A nucleic acid comprising any one of the following polymorphisms:

- the nucleic acid of SEQ ID NO.1 with A at position 344 as defined by the positions in SEQ ID NO.1; the nucleic acid of SEQ ID NO.1 with six Gs at positions 621-626 as defined by the 30 positions in SEQ ID NO.1; the nucleic acid of SEQ ID NO.1 with six Gs at positions 793-798 as defined by the positions in SEQ ID NO.1; the nucleic acid of SEQ ID NO.1 with T at position 908 as defined by the positions in SEQ ID NO.1; the nucleic acid of SEQ ID NO.1 with C at position 1136 as defined by the positions in SEQ ID NO.1; the nucleic acid of SEQ

ID NO.1 with C at position 1160 as defined by the positions in SEQ ID NO.1; the nucleic acid of SEQ ID NO.1 with A at position 1189 as defined by the positions in SEQ ID NO.1; the nucleic acid of SEQ ID NO.1 with G at position 1458 as defined by the positions in SEQ ID NO.1; the nucleic acid of SEQ ID NO.1 with G at position 1656 as defined by the positions

5 SEQ ID NO.1; the nucleic acid of SEQ ID NO.1 with C at position 2448 as defined by the positions in SEQ ID NO.1; the nucleic acid of SEQ ID NO.1 with A at position 2531 as defined by the positions in SEQ ID NO.1; the nucleic acid of SEQ ID NO.1 with T at position 3348 as defined by the positions in SEQ ID NO.1; the nucleic acid of SEQ ID NO.1 with G at position 3432 as defined by the positions in SEQ ID NO.1; the nucleic acid of SEQ ID NO.1

10 with A at position 3622 as defined by the positions in SEQ ID NO.1; or a complementary strand thereof or an antisense sequence thereto or a fragment thereof of at least 17 bases comprising at least one polymorphism.

11 10. An allele specific primer or probe capable of detecting an EP1-R gene polymorphism

15 at one or more of positions 344, 621-627, 793-799, 908, 1136, 1160, 1189, 1458, 1656, 2448, 2531, 3348, 3432, and 3622 in the EP1-R gene as defined by the positions in SEQ ID NO.1.

12 11. A diagnostic kit comprising one or more diagnostic primer(s) and/or probes(s) as defined in claim 10.

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13 12. A method of treating a human in need of treatment with an EP1-R drug in which the method comprises:

i) diagnosis of a polymorphism in the EP1-R gene in the human, which diagnosis comprises determining the sequence of the nucleic acid at one or more of positions 344, 621-627, 793-799, 908, 1136, 1160, 1189, 1458, 1656, 2448, 2531, 3348, 3432 and 3622 in the EP1-R gene as defined by the positions in SEQ ID NO.1, and determining the status of the human by reference to polymorphism in the EP1-R gene; and

ii) administering an effective amount of a EP1-R drug.

30 14 13. Use of an EP1-R drug in the preparation of a medicament for treating an EP1-R mediated disease in a human diagnosed as having a polymorphism at one or more of positions

344, 621-627, 793-799, 908, 1136, 1160, 1189, 1458, 1656, 2448, 2531, 3348, 3432 and 3622 in the EP1-R gene as defined by the positions in SEQ ID NO.1.

14. A pharmaceutical pack comprising an EP1-R drug and instructions for administration
5 of the drug to humans diagnostically tested for a polymorphism at one or more of positions
344, 621-627, 793-799, 908, 1136, 1160, 1189, 1458, 1656, 2448, 2531, 3348, 3432 and 3622
in the EP1-R gene as defined by the positions in SEQ ID NO.1.

15. A computer readable medium having stored thereon a nucleic acid sequence
10 comprising at least 17, preferably at least 20 consecutive bases of the EP1-R gene sequence,
which sequence includes at least one of the polymorphisms at positions: 344, 621-627, 793-
799, 908, 1136, 1160, 1189, 1458, 1656, 2448, 2531, 3348, 3432 and 3622 in the EP1-R gene
as defined by the positions in SEQ ID NO.1.

15. A method for performing sequence identification, said method comprising the steps of
providing a nucleic acid sequence comprising at least 20 consecutive bases of the EP1-R gene
sequence, which sequence includes at least one of the polymorphisms at positions: 344, 621-
627, 793-799, 908, 1136, 1160, 1189, 1458, 1656, 2448, 2531, 3348, 3432 and 3622 in the
EP1-R gene as defined by the positions in SEQ ID NO.1, in a computer readable medium; and
20 comparing said nucleic acid sequence to at least one other nucleic acid sequence to identify
identity.

16. A purified allelic variant of the human EP1-R polypeptide having a proline at position
126 and/or a threonine at position 154 or a fragment thereof comprising at least 10 amino
25 acids provided that the fragment comprises the allelic variant at position 126 and/or position
154.

17. An antibody specific for an allelic variant of human EP1-R polypeptide having a
proline at position 126 and/or a threonine at position 154 or a fragment thereof comprising at
30 least 10 amino acids provided that the fragment comprises the allelic variants at position 126
and /or position 154.